Original Article

# Review of Congenital Anomalies of Ear, Nose, and Throat in a Resource-Challenged Facility

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## Abstract

**Introduction:** The complex pattern of development of the branchial arches predisposes to a myriad of congenital anomalies of the head-and-neck region. The ear, nose and throat (ENT) are by no means spared. The birth of a child with congenital anomaly elicits emotion, anxiety, the feeling of guilt, and dejection in the parents, especially the mother. They are worried and anxiously seek for the explanation which the attending doctor who is ill-equipped to venture into it. The etiological factors are many and vary. ENT anomalies are often associated with malformations of other organs and systems and should be searched for and detected. Any treatment offered should aim at restoring normal or near-normal appearance and function and allay the fears and apprehensions of the parents. **Aim:** The aim of the study was to review and document the incidence, variety, and presentation of ENT congenital anomalies seen and treated in the Otolaryngology Department of our institution and formulate a baseline for the future. **Materials and Methods:** It was a retrospective review of ENT anomalies handled in our hospital from January 2015 to December 2019. The case notes of the eligible cases were retrieved and the relevant data were extracted. The data collected were analysed with descriptive statistics and presented in tables and prose as deemed fit. **Ethical Consideration:** The study protocol was reviewed and approved by the hospital records department. **Results:** Forty-six cases were studied, 24 males and 22 females. Their ages ranged from 0.019 years (one week) to 55 years, average of 7.84 ± 10.38, 95% confidence interval of 4.75682–10.92318. There was no significant difference in the ages of the males and females $P = 0.8809$, $t = 0.1507$, dt = 44. Fourteen different malformations were detected most common of which was deafness 15 (32.61%), followed by thyroglossal cyst 7 (15.22%) with 5 different anomalies coming last with 2.17% each. **Conclusion:** ENT malformations are common in our locality. Adequate history and examination with appropriate investigations will help get the diagnosis and associated conditions. Multi-disciplinary approach to management will offer a better outcome. Parents and caregivers need to be properly counselled.

**Keywords:** Anomaly, congenital, ear, nose, review, throat

## Introduction

The congenital anomalies of the ear, nose, and throat (ENT) are no doubt part of the craniofacial anomalies; which literally encompass all congenital deformities of the head that interfere with physical and mental well-being.[1] The complex pattern of development of the branchial arches predisposes to many congenital malformations in the ear, nose, and throat. A child born with any congenital anomaly elicits obvious feelings of anxiety and palpable guilt in the parents. The very obvious deformities of the ear, nose, and cranium cannot be hidden from the world, especially in a newborn. For instance, the nose, which is the most outstanding feature of the face, many parents will observe and comment on minor abnormalities even when these are barely visible and causing no functional disability. More major aberrations of the external nose may induce considerable parental dismay and even, occasionally give rise to the rejection of the baby. The parents of the affected child ask their doctor for an explanation and the doctor usually cannot give a reason why the child is the 1 in 20,000 who develops a congenital anomaly. Many causes ranging from hereditary to viral infections and maternal nutritional deficiencies have been implicated. When a child presents with

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an inborn deformity, one must look for associated anomalies of other organs and systems because ear, nose, and throat malformations may be associated with cardiovascular or gastrointestinal malformations.

The goal of treatment offered should be to give if not a normal, at least a near-normal appearance and functional profile.

Together with the management of the child, the parents should be counselled and advised douncing their emotions and fears and outlining a realistic expectancy regarding cure.

The aim of the study was to review and document the incidence, variety, and presentation of congenital anomalies of the ear, nose, and throat seen in the Otolaryngology Department of our institution. It is hoped that the outcome of the study will form a baseline for future expectations and direct the course for intervention.

## Materials and Methods

The study was a retrospective review done in the Department of Otolaryngology, Head and Neck Surgery of our hospital during the period January 2015–December 2019. All patients with inborn malformations of ENT treated in our hospital by the otolaryngologist and other support units were included in the study. Their case notes were retrieved from the ward, clinic, theatre, accident, emergency department, and intensive care unit for the relevant data. The variables studied included patients' age, gender, diagnosis, interventions, and outcome.

The data generated were analysed using descriptive statistics and presented in tables, and prose or descriptive forms as appropriate.

### Ethical consideration

The study protocol was reviewed and approved by the hospital medical records department.

## Results

Forty-six cases with congenital anomalies of the ear, nose, and throat were studied. Out of the 46 patients, 24 (52.2%) were males and 22 (47.8%) were females with male:female ratio of 1.1:1.0. The mean average age and standard deviation of the patients were 7.84 ± 10.38 with 95% confidence interval (CI) of 4.75682–10.92318. The ages of the males ranged from 0.019 years (one week) to 55 years, median three years with a mean of 8.06 ± 12.40, 95% CI of 2.82705–13.29953 while that of females spanned from 0.038 years (two weeks) to 30 years, median three years with a mean of 7.60 ± 7.90; 95% CI of 4.09237–11.10017. There was no significant difference in the ages of the males and females studied, $P = 0.8809$, $t = 0.1507$, dt = 44.

Of the 46 cases studied, 35 cases were presented in the first decade of life. There were 3 newborns and 11 infants; 7 in the second decade of life and 4 cases over the age of 20 years.

Fourteen different anomalies were detected in the study ranging from deafness through thyroglossal cyst to haemagioma that was seen as an oval mass on the palate among others [Table 1].

## Discussion

A fairly good number of conditions were detected that mirrored what is generally known. Both males and females were almost equally affected. A close look at the most common conditions detected may suffice and give further insight to the conditions.

### Deafness

The most common anomaly detected in the study was congenital deafness. It accounted for 32.61% of the cases [Table 1]. Their audiograms revealed a severe degree of sensorineural hearing loss. Two of the cases were described as deaf-mutism. It was not clear in the case notes the cause of the deafness. The possibility of prenatal exposure to herbal medicine and anoxia from prolonged labor may not be ruled out as most of the cases were referred from outside. Newton[2] in his article described the problems associated with assigning a cause to hearing loss for many children.

### Thyroglossal cyst

This was the second most common anomaly (15.22%) in the study. Out of the 7 cases, 3 were males and 4 females; 3 were in the first decade of life, 1 and 3 above the second decade. In general, thyroglossal cysts occur equally in men and women and are usually noted in childhood, but may present at any age. The cyst can occur at any point along the path of descent of the primitive thyroid gland the tract from the base of the tongue to the thyroid isthmus, the most common site being just above or just below the hyoid bone. Most cysts are midline, but those at the level of the thyroid cartilage may be pushed to one or other side, usually the left. On examination, it is freely mobile and may transilluminate. It rises on swallowing because of its attachment to the hyoid bone through the thyroglossal tract and

**Table 1: The frequency of the various anomalies detected and percentage ranking**

| Anomalies | *n* (%) |
|---|---|
| Deafness | 15 (32.61) |
| Thyroglossal cyst | 7 (15.22) |
| Choanal atresia | 4 (8.70) |
| Preauricular sinus | 4 (8.70) |
| Laryngomalacia | 3 (6.52) |
| Aural (meatal) atresia | 2 (4.35) |
| Speech disorder | 2 (4.35) |
| Preauricular cyst | 2 (4.35) |
| Branchial cyst | 2 (4.35) |
| Crouzon syndrome | 1 (2.17) |
| Dermoid cyst | 1 (2.17) |
| Cystic hygroma | 1 (2.17) |
| Ankyloglossia | 1 (2.17) |
| Haemagioma | 1 (2.17) |
| Total | 46 (100.00) |

rises on protrusion of the tongue due its attachment to the base of the tongue through the thyroglossal tract. No other midline neck swelling rises on tongue protrusion, so this physical sign is virtually pathognomonic for the diagnosis. Some patients present with acute infection and abscess formation which may result in a sinus or fistula with an intermittent discharge of clear glary mucus. Such a sinus or fistula is always acquired and may follow inadequate surgical excision. When the cyst is cosmetically unacceptable or is the site of recurrent infection and fistula formation, it is surgically removed. Gross and Connerley[3] have argued that small symptomless cysts maybe left *in situ*. However, many cysts do eventually become the site of recurrent inflammation and, very occasionally, a carcinoma may develop within the cyst. In most cases, therefore, surgical excision is the treatment of choice. A preoperative thyroid scan should be performed and this will usually show a normal thyroid gland in the normal position. In 1920, Sistrunk[4] described his technique for the removal of thyroglossal cysts. He had found that the simple excision of the cyst was often followed by further cyst formation or a chronically discharging sinus at the operation site. He posited that those recurrences could be avoided by removing the whole of the thyroglossal tract. To achieve complete removal, he advised excision of the central part of the body of the hyoid bone and core of tongue muscle up to the foramen cecum. Using this technique on 270 patients at Mayo clinic, Brown and Judd[5] were able to reduce the previously high recurrence rate to only 4%. In all cases, a core of the tongue tissue comprising parts of geniohyoid and genioglossus should be excised to ensure that all tract remnants including accessory tracts are removed.[6,7] Thyroid carcinomas are occasionally found in thyroid cysts. Page *et al*.[8] reviewed the literature and found 656 such cases, all of which were papillary carcinomas. Most had presented as simple cysts and were diagnosed only on histological examination of the tissue. Treatment is adequate surgical excision, followed by suppressive doses of thyroid hormones.

## Choanal atresia

Four (8.70%) cases of choanal atresia were included in the study; occurred equally in both males and females. Three were unilateral and one bilateral and presented at one week (0.019 years). The chonae are the posterior apertures of the nose.[9] Atresia of the choanae was first described by Roederer in 1775[10] It is one of the more frequently observed congenital abnormalities of the nose, although its true incident is uncertain. The most consistently quoted figure is 1/8000 live births, but this is probably an underestimate. Bilateral choanal atresia is likely to have been, and may still be, a frequently unrecognized cause of death in the neonatal period. Contrary to our finding in the study, females appear to be affected about twice as often as males and the condition may be unilateral or bilateral in the proportion 3:2.[11] Uncertainty exists about the embryological structure involved in choanal atresia. Kaplan, Sprinkle and Sporck[11,12] subscribed to the theory that the anomaly may arise as a result of the persistence of the buccopharyngeal membrane. It has also been suggested that the aberration is due to the failure of the bucconasal membrane to undergo involution.[12] Choanal atresia may be bony (90%) or membranous (10%), occasionally mixed in a negligible number of cases, and is generally sited just in front of the posterior end of the nasal septum. In most instances, where the atretic plate is of bony type, it is thin and easily perforated. In the newborn, choanal atresia can present as an emergency with virtually total apnea until the mouth is opened and an airway inserted. In an emergency situation, an airway can be taped in place to avoid intubation. Often unilateral choanal atresia is not diagnosed until later in childhood or even adult life. Even though choanal atresia may be found as an isolated anomaly, about 60%–70% of cases are associated with one or more other congenital defects.[11,13] Apart from random associations, choanal atresia has been linked to a number of specific defects – the so-called CHARGE (coloboma, heart disease, atresia of choanae, retarded growth, genital hypoplasia, and ear abnormalities) association.[14] The diagnosis is made by mirror test, inability to pass a soft catheter or probe through the nose into the nasopharynx, air blown into the nostril through a closely fitting tube is not heard to enter the nasopharynx or the use of a stethoscope from which the bell has been removed, the open end of the tube being held over each nostril in turn and the presence or absence of air blast noted. Confirmation can be made by choanography-contrast radiography where a contrast medium (radio-opaque medium) is held up at the choana. Flexible nasopharyngoscopy/diagnostic nasal endoscopy may also be used. The current investigation of choice, however, is computerised tomography to delineate the nature and thickness of the obstruction and the actual structure involved.[15] However, attention has been drawn to the potentially misleading effects of mucus within the nasal cavity; stressing the need for careful preparation, including the use of vasoconstrictive drops and nasal suctioning if high-quality images are to be obtained.[16] Treatment is aimed at the creation of patent airways. This is achieved through one of four approaches, namely transnasal, transpalatal, transseptal, and transantral. Of these, only the first two are in common use. In all surgery of this region, the use of an operating microscope with a teaching arm or camera fitted to it and conveyed on a monitor not only provides much better illumination but makes the operation easier and safer. Some authors are of the view that endoscopic transnasal repair provides better visualisation and the ability to perform more exact surgery.[17,18]

## Preauricular sinus

There were 4 (8.70%) cases of preauricular sinus in the study, 3 females and 1 male; all presented in the first decade of life asymptomatically. They were all unilateral, 3 right and 1 left congenital sinuses composed of blind tracks lined by squamous epithelium are commonly found in the preauricular region along the ascending crus of the helix. Others may open along a line extending from the lower border of the helix to the angle of the mouth. Collaural fistulae open superiorly in the floor of the external auditory meatus and inferiorly at the anterior border of the sternomastoid behind the angle of the jaw. Many authors

have found an association between preauricular sinuses, branchial fistulae, and deafness.[19,20] Congenital sinuses and fistulae do not require treatment unless they cause symptoms. Occasionally, they become infected leading to a persistent discharge and sometimes to abscess formation. At this point, surgical intervention is indicated which is complete excision. During the procedure, adequate care is taken as the tract often extends deep into the soft tissues and can be intimately related to branches of the facial nerve. Before excision, the tract is injected with gentian violet dye to aid its identification and complete removal.

### Laryngomalacia

Three cases, two males presented at three months (0.25 years) and one female at one year. The condition may appear within an hour or two of birth and is the most common cause of stridor in infants. Stridor is the only symptom and appears at or soon after birth. It is croaking in character and mainly inspiratory in phase. It is reduced by rest, and responds to changes in posture but worsened by exertion. It disappears between the second and fifth years of life. Cyanosis is rare and the voice is unchanged. Diagnosis can be made by a careful history and examination. Inspiratory stridor without hoarseness is always suggestive when occurring at or soon after birth. Flexible fiber-optic examination, or direct laryngoscopy, will distinguish it from other conditions of the larynx. Reassurance is usually all that is necessary in this condition. Tracheostomy as often requested by physicians and other units is virtually unknown for laryngomalacia and, if considered, an associated vocal cord paralysis should be suspected and carefully excluded. It should be avoided whenever possible as it carries a 5%–10% mortality. The other less common conditions identified were addressed on their own merit.

## Conclusion

Congenital deformities of the ear, nose, and throat are fairly common in our environment.

Detailed history with a painstaking and thorough examination and appropriate investigation would be needed to arrive at the diagnosis and discover the associated anomalies. Many congenital malformations would be better managed by a multidisciplinary approach in dedicated centres where necessary otolaryngological, radiological, maxillofacial, neurosurgical, and other skills are readily available. Parents of the affected individuals should be adequately counseled and given a realistic expectation of the outcome of any intended intervention proposed.

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### Conflicts of interest

There are no conflicts of interest.

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